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(FILE 'HOME' ENTERED AT 18:20:59 ON 09 FEB 2003)

FILE 'HCAPLUS' ENTERED AT 18:21:11 ON 09 FEB 2003

E SITOSTEROLEMIA SUSCEPTIBILITY GENE/CT

L1 1 S SITOSTEROLEMIA SUSCEPTIBILITY GENE

E ABC TRANSPORTER/CT

E E4+ALL

E E2

E E3+ALL

L2 417 S E3

L3 75 S SITOSTEROLEM?

L4 1 S L2 (L) L3

=> e e3+all

E1' 609132 BT2 Proteins/CT

E2 19180 BT1 Transport proteins/CT

E3 --> Transport proteins (L) ABC (ATP-binding cassette)
transporters/CT

E4 OLD Proteins, specific or class (L) ABC (ATP-binding
cassette-contg.)/CT

E5 OLD Transport proteins (L) ABC (ATP-binding
cassette-contg.)/CT

E6 UF ABC proteins/CT

E7 UF ABC transporters/CT

E8 UF ATP-binding cassette transporters/CT

E9 UF ATP-binding cassette-contg. proteins/CT

=> d ibib ab 1

L4 ANSWER 1 OF 1 HCAPLUS COPYRIGHT 2003 ACS

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TITLE: Novel donor splice site mutation of ABCG5 gene in sitosterolemia

AUTHOR(S): Lam, Ching-Wan; Cheng, Anna Wai-Fun; Tong, Sui-Fan; Chan, Yan-Wo

CORPORATE SOURCE: Department of Medicine, Brigham and Women's Hospital, Boston, MA, 02115, USA

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CODEN: MGMEFF; ISSN: 1096-7192

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AB In a patient with sitosterolemia, we found two different mutations of the ATP-binding cassette, subfamily G, member 5 (ABCG5) gene. The first is a missense mutation that changes the amino acid residue at position 419 from arginine to histidine, i.e., R419H. The second is a novel splicing mutation affecting the invariant guanine at the first base of the donor splice site of intron 12, i.e., IVS12+1G A. The father of the patient is heterozygous for the missense mutation, and the mother is heterozygous for the splicing mutation. No mutations were found in the sister of the patient. Up until now, the missense mutation has only been found in Japanese patients with sitosterolemia. We believe that R419H in our Chinese patient may have the same origin as the mutation in the Japanese patients with sitosterolemia. (c) 2002 Academic Press.

REFERENCE COUNT: 4 THERE ARE 4 CITED REFERENCES AVAILABLE FOR THIS RECORD. ALL CITATIONS AVAILABLE IN THE RE FORMAT